[Medicinski fakultet u Rijeci]

Curriculum
2022/2023

[Za kolegij]

Medical genetics

Study program:	**Medical Studies in English (R)** [Sveučilišni integrirani prijediplomski i diplomski studij]
Department:	**[Katedra za medicinsku biologiju i genetiku]**
Course coordinator:	**izv. prof. dr. sc. Pereza Nina, dr. med.**
Year of study:	**5**
ECTS:	**3**
Incentive ECTS:	**0 (0.00%)**
Foreign language:	**Possibility of teaching in a foreign language**

Course information:

Course Medical genetics is a mandatory course based on clinical reasoning in the fifth year of the University integrated undergraduate and graduate study of Medicine in English, and consists of 18 hours of lectures, 14 hours of seminars and 13 hourse of practicals, with a total of 45 hours (3 ECTS). The aim of the course is to describe and explain the comprehensive approach to a patient with a genetic disease or disorder or an increased risk for them, so that future physicians can apply the acquired knowledge, skills and attitudes in their own clinical practice.

COURSE LEARNING OUTCOMES:

Learning outcomes are determined and derived in accordance with the basic standards for the development of genetics competencies for health professionals in Europe, specifically physicians who are not specialists in medical genetics. These competencies are contained in the document Core competencies in genetics for health professionals in Europe (EuroGentest Project, https://www.eshg.org/index.php?id=139), which was accepted and approved by the Education Committee of the European society for human genetics in 2008.

After passing the exam, the student will be able to:

A. COGNITIVE DOMAIN – KNOWLEDGE

1. list and distinguish the types of genetic disorders as causes of diseases and medical conditions

2. list and compare the types and outcomes of genetic testing according to groups of indications

3. argue the advantages and limitations of genetic tests and methods used

4. calculate the recurrence risk of genetic disorders

5. distinguish categories of developmental anomalies and associate them to the corresponding causes

6. use the standardised terminology when describing dysmorphic features

7. distinguish the effects of gene variability on therapeutic outcome

B.PSYCHOMOTORIC DOMAIN – SKILLS

1. recognise patients with a genetic disease or disorder, as well as increased risk for them

2. choose the appropriate method of genetic testing according to indication and genetic cause of a disease or medical condition

3. basically interpret the findings of genetic testing

4. organise genetic care by referring patients to medical genetics specialists, as well as other appropriate specialists and experts

5. apply basic communication skills in the transmission of genetic information

6. search diagnostic and educational databases of genetic diseases and disorders (OMIM, GeneReviews, Orphanet, Human Phenotype Ontology, Face2Gene, PharmGKB)

C. AFFECTIVE DOMAIN – VALUES AND ATTITUDES*

1. judge and identify the importance of making a genetic diagnosis

2. adopt the specificities of the approach to patients in medical genetics in relation to other medical specialisations

3. respect the importance of emotional, psychological, social and material consequences of genetic testing findings

4. apply the acquired knowledge and skills in providing the basic level of psychological and social support to patients and families affected by genetic disease or disorder

5. recognize the need for lifelong learning in medical genetics

**The course Medical Genetics places special emphasis on the development of the affective domain, as it is crucial that, in addition to the mentioned specific knowledge and skills, future physicians develop awareness for the multidimensionality of genetic diseases and disorders. They not only encompass the cellular level in a patient but are also reflected in all other, higher aspects of the patient's life, as well as those of their blood relatives and offspring. Genetics permeates all branches of medicine and all of humanity, from the structure and function of nuclear or mitochondrial DNA, a single cell, tissue, integrated organs, all the way to the psychological superstructure ofthe human body. As such, medical genetics is one of the most complex medical specialisations, as it requires the integration of all knowledge about the structuring and functioning of the human body and spirit.*

COURSE CONTENT:

With the aim of purposeful and meaningful realisation of learning outcomes, classes are organized in five large thematic units that answer specific questions related to approach to a patient with a genetic disease or disorder or increased risk for them:

1. INDICATIONS FOR GENETIC TESTING

(How to recognize a person with a genetic disorder?)

P1	How to distinguish between genetic and non-genetic aetiology of diseases and medical conditions?
L2	Types of genetic testing according to indications
L3	The path to genetic diagnosis: care organization and diagnostic tools in medical genetics

2. TYPES OF GENETIC DISORDERS

(How to choose the proper method of genetic testing and interpret the results at a basic level?)

Disorders within genes	
L4	Spectrum of phenomena in autosomal dominant monogenic diseases
L5	Autosomal recessive monogenic diseases: from patient to carrier
L6	X-linked monogenic diseases and other rare types of inheritance
S1	One disease – one monogenic cause
S2	One disease – several possible monogenic causes: locus heterogeneity
S3	One disease – several possible different causes: complex diseases
Disorders of one part or entire chromosome	
L7	Deviations from the normal chromosome number
L8	Balanced structural genomic rearrangements: translocations and inversions
L9	Unbalanced structural genomic rearrangements: genomic disorders
S4	Reproductive genetics I: fertility disorders and pregnancy complications
L10	Prenatal screening and diagnostics of chromosomopathies
S5	Reproductive genetics II: prenatal diagnostics
Disorders of the (epi)genome	

L11	Uniparental disomy and epimutations

3. THINKING IN PATTERNS OF CLINICAL FEATURES IN WIDE DIFFERENTIAL DIAGNOSIS

(How to direct genetic testing in persons with multiple congenital anomalies with or without intellectual disabilities?)

L12	Classifications and causes of congenital anomalies
L13	Basics of clinical dysmorphology
S6	Genetics of mental development: developmental delay, intellectual disabillity and autism spectrum disorders
P2	How to recognize patterns of multiple congenital anomalies?

4. PERSONALIZED MEDICINE

(How to approach each patient individually?)

L14	Practical pharmacogenomics
L15	Gene therapy in clinical practice
S7	Interpretation of results in pharmacogenomics
P3	How to organize comprehensive patient care?

5. COMMUNICATION SKILLS IN MEDICAL GENETICS

(How to convey genetic information to a patient?)

L16	Specific challenges in genet(h)ic counseling
L17	Basic communication skills in medical genetics
P4	Simulation of clinical reasoning in medical genetics
P5	How to convey genetic information to a patient?

The concept of individual forms of teaching is based on the following:

Lectures – From clinical picture to genetic causes and diagnostic methods

Seminars - From recognising the indication to interpreting the findings of genetic testing

Practicals - Implementation of acquired competencies in one's own clinical practice

COURSE CONCEPT:

Course Medical genetics is a course of clinical reasoning, the foundations of which are based on stories about patients, from whom it all begins, specifically on those examples of genetic diseases or disorders that physicians most often encounter after graduation (examples from cardiogenetics, neurogenetics, oncogenetics, pediatric genetics, gynecology, etc.). With the aim of acquiring specific knowledge, skills and values / attitudes in medical genetics, all forms of teaching (lectures, seminars and practicals) are conducted through active learning techniques, encouraging the development of open, analytical and critical thinking. All material will be presented through case analyses (case-based learning), problem-based and experiential learning in a way that reflects the authentic form of action in clinical practice, in which the physician first meets the patient, not their molecular structure. This approach to learning and teaching, reverse to the classical way, encourages simpler and more meaningful mastery and application of basic theoretical knowledge, as well as thinking about the patient in patterns of clinical features in making a working / clinical diagnosis and directing genetic testing. Therefore, lectures, seminars and exercises will

be held in an interactive environment.

List of assigned reading:

1. Pereza N. Handbook with case reports in Medical genetics. First edition. 2021. Rijeka: University in Rijeka, Faculty of Medicine, Department of medical biology and genetics. Available at: https://repository.medri.uniri.hr/en/islandora/object/medri%3A4811

2. Materials from lectures

List of optional reading:

1. Turnpenny P, Ellard S. Emery's elements of medical genetics. 14. ed. 2011. Philadelphia: Elsevier/Churchill Livingstone.

2. Read A, Donnai D. New Clinical Genetics. Third edition. 2015. Banbury, UK: Scion Publishing Limited.

Curriculum:

Student obligations:

All information regarding the course, as well as materials from the lectures will be available on the Merlin elearning platform. Students should visit the mentioned platform regularly in order to be informed in a timely manner of any facts or changes concerning the course. Furthermore, students should regularly fulfill the obligations related to course attendance and active participation in classes.

COURSE ATTENDANCE:

Classes are organized according to the schedule published on the Merlin e-learning platform and INP application. Attendance of lectures, seminars, practicals and midterm exams is mandatory and attendance records are kept separately for each student. All of the previously mentioned types of classes start at the exact time according to the specified schedule and being late is treated as absence from class. Entries / exits during classes are not allowed. A student may justifiably miss up to 30% of the hours provided separately for practicals, seminars and lectures, solely for health reasons, which is confirmed by a medical certificate (including absences from midterm exams). If a student is unjustifiably absent from more than 30% of class hours for each class type (5 hours of lectures, 4 hours of seminars, 4 hours of exercises), the student cannot continue to attend the course and loses the opportunity to take the final exam (0 ECTS, grade F).

ACTIVE PARTICIPATION IN CLASSES:

Since the course is implemented through forms of active learning, students must have and use the Handbook with case reports from Medical Genetics in all forms of classes (lectures, seminars and practicals) (either in electronic or printed version). During certain classes, students will independently use information technology, including active search and use of genetic electronic databases freely available on the Internet. Therefore, it is recommended to use smart phones in contact form, especially during seminars and practicals.

Exam (exam taking, description of the written/oral/practical part of the exam, point distribution, grading criteria):

Student assessment is carried out according to the current regulations at the University of Rijeka, and according to the Ordinance on student grading at the Medical Faculty in Rijeka (adopted by the Faculty Assembly of the Medical Faculty in Rijeka). Assessment is carried out using ECTS grading system (% / A-F) and a numerical grading system (1-5). Student progress will be evaluated and graded during classes and at the final exam. Out of a total of 100 credits, the student can achieve a maximum of 70 credits (70%) during the course in two written midterm exams, and a maximum of 30 credits (30%) in the final, structured oral exam.

I. DURING COURSE - MIDTERM EXAMS (TOTAL MAXIMUM OF 70 CREDITS):

During the course, the acquired knowledge from lectures, seminars and practicals will be assessed by two midterm exams in the form of a written test with multiple choice questions (Midterm exams I and II). At each midterm exam, the criterion for obtaining credits is at least 50% of correctly answered questions. Passed midterm exams are not transferable and are valid for the current academic year. Midterm exams I and II will be conducted onsite at the Faculty of Medicine in Rijeka.

Midterm exam I includes teaching units L2-P10, S1-5 and P1, has 40 questions and carries up to 40 credits. Writing time is 50 minutes. The number of correctly answered questions is converted into credits as follows:

Number of correct answers	0-19	20	21	22	23	24	25	26	27	28	29	30	31	32	33	34	35	36	37	38	39	40
Credits	0	20	21	22	23	24	25	26	27	28	29	30	31	32	33	34	35	36	37	38	39	40

Midterm exam II includes teaching units L11-17, S6-7 and P2-5, has 30 questions and carries up to 30 credits. Writing time is 40 minutes. The number of correctly resolved questions is converted into credits as follows:

Number of correct answers	0-14	15	16	17	18	19	20	21	22	23	24	25	26	27	28	29	30
Credits	0	15	16	17	18	19	20	21	22	23	24	25	26	27	28	29	30

Midterm repetition

Midterm exam repetitions can be accessed by students who:

• did not collect 35 credits during the classes, i.e. did not pass the midterm exam I and/or II, and/or

• were justifiably absent during classes during the midterm I and/or II exams (due to illness, with a medical certificate), and/or

• have passed the midterm exam I and/or II, but are not satisfied with the achieved credits

In case of midterm I and/or II repetition, the final credits are those that the student achieves on the repeated midterm exam. Midterm exam repetitions can be accessed only once for each midterm exam. Repetitions are held live after the completion of regular classes in two terms, in each of which only one midterm is retaken:

Midterm I repetition – in agreement with students ----- Midterm II repetition – in agreement with students

The students can apply for repetitions of midterm exams via the e-mail address of the Secretary of the Department of medical biology and genetics no later than two days before the repetition of the midterm exams.

II. FINAL EXAM (TOTAL MAXIMUM OF 30 EVALUATION POINTS):

The final exam cannot be accessed by students who:

• finally achieve less than 35 credits after having taken the repetition of the midterm exam I and/or II, and / or

• have 30% or more of unjustified absences from classes.

Such a student is graded with an F (unsuccessful), cannot obtain ECTS credits or take the final exam, i.e. must re-enroll the course in the following academic year.

The final exam can be accessed by

• achieved ≥35 credits (50% or more of the possible 70 credits) during classes, and

• do not have more than 30% of justified absences from classes.

The final exam is a structured oral exam in the form of a patient management problem that will be held onsite at the Faculty of Medicine in Rijeka and consists of four patient cases that will examine the integration of knowledge, skills and values / attitudes (monogenic disorder, chromosomal disorder, dysmorphic syndrome, pharmacogenomics). The final exam is passed if the student achieves 15 out of 30 credits.

Success in the structured oral exam is evaluated and converted into credits in the following way :

	Credits	
Grade	**Case 1-3**	**Case 4**
the answer meets the minimum criteria	4	3
averagely good answer	5	4
very good answer	6-7	5
excellent answer	8	6

III. FINAL GRADE:

The final grade is the sum of the credits collected during the course and at the final exam. Assessment within the ECTS system is carried out according to the achieved final success in the following way:

Percentage of assessed credits	**ECTS grade**	**Numerical grade**
90 - 100	A	excellent (5)
75 - 89,9	B	very good (4)
60 – 74,9	C	good (3)
50 - 59,9	D	sufficient (2)
0 - 49,9	F	insufficient (1)

Other notes (related to the course) important for students:

COMMUNICATION WITH TEACHERS:

Teachers are available daily during working hours via e - mail addresses (available on the webpage of the Faculty of Medicine in Rijeka) for all questions concerning the course. Consultations are possible by appointment and can be conducted live or through the online platform MS Teams.

ACADEMIC INTEGRITY:

It is expected that the teacher will respect the Code of Ethics of the University of Rijeka, and the students the Code of Ethics for students of the University of Rijeka .

COURSE HOURS 2022/2023

Medical genetics

List of lectures, seminars and practicals:

EXAM DATES (final exam):
